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Case Report

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Startle syndrome in a neonate-a case report

Abstract

Startle Syndrome/Hyperekplexia are a rare, neurological disorder that may affect newborns, children and adults. The disease is caused by multiple genetic mutations. The early manifestations include abnormal responses to unexpected auditory, visual, and so esthetic stimuli such as sustained tonic spasm, exaggerated startle response and fetal posture with flexion of forearm and legs, clenched fists, and anxious stare. Spasms may be severe enough to cause apnea, bradycardia and death. These movements must be distinguished from startle epilepsy. Diagnosis is based on the clinical signs, molecular genetic testing and electrophysiology. Prognosis is variable. Diazepam and clonazepam are considered the drugs of choice. Forced flexion of the head and legs towards the trunk (Vigevano maneuver) can be life saving when prolonged stiffness impedes respiration. A timely diagnosis in cases of hyperekplexia is crucial because affected neonates are at risk of sudden death from apnea. A neonatal case is presented below.

Keywords: startle syndrome, hyperekplexia, clonazepam, vigevano maneuver, genetic counseling

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Introduction

Hyperekplexia is characterized by generalized stiffness immediately after birth that normalizes during the first years of life; excessive startle reflex to unexpected particularly auditory stimuli; and a short period of generalized stiffness following the startle response during which voluntary movements are impossible.[1] The disorder was first described in 1958 by Kirstein and Silverskiold, who reported a family with 'drop seizures'.[2] The prevalence rate of Hyperekplexia is <1/1000000.[3] The disease is caused by multiple genetic mutations. Specifically, the mutations affect the movement of glycine between cells. The new findings firmly establish mutations in the GlyT2 gene as a major cause of the disease.[4]

Case report

A 26 day old male neonate born to non consanguineous healthy parents was admitted with the sole complaint of episodic abnormal movements, posturing and startle response to sound or touch since birth. Baby was born at term by normal vaginal delivery following an uneventful pregnancy. Apgar scores were 8 at the first minute and 9 at the fifth minute respectively; no resuscitation was required. The birth weight was 3100g. Head circumference was normal. Baby was shifted to NICU as he developed hypoglycemia and hypocalcemia with seizure. Diazepam had been prescribed for the suspicion of neonatal seizures. EEG displayed no changes. On day 3, baby developed two episodes of stiffness with staring. Managed with diazepam. Baby was on formula feed till day 5 and was discharged on day 7 as baby sucked well from the breast and displayed no abnormalities. After 19 days in home at the time of routine bath, baby cried, developed stiffness, posturing and breath holding spell. Shifted immediately to tertiary care centre. Routine investigations were normal. Video EEG displayed no abnormalities. Tone was symmetrically increased and ankle clonus was present. Deep tendon reflexes were increased. Diagnosis of hyperekplexia was made and baby was on syrup Levipil, tab Clonazepam. Baby demonstrated improved tone and was discharged with medications to continue along with physical therapy. At 6 month follow-up the child has shown continued improvement in the form of increased spontaneous movements, decreased startle and achievement of most of the normal milestones however head turn and gaze deviation is persisting.

Discussion

Rigidity decreases spontaneously in toddler age group when increase in spontaneous activity occurs. However, in few cases mild stiffness reappears in adolescence or adult life and spasms may again be provoked by startle. Such episodes are a source of injury. Unpredictable sudden falls or undue startle may make outdoor activities both dangerous and embarrassing resulting in considerable social dysfunction. Delayed motor milestones are present while cognitive function normally remains unaffected, though low intelligence has been noted in some studies.[5] Clonazepam which enhances GA-BA-gated chloride channel functions and presumably compensates for defective glycine gated chloride channel functions has been considered the first choice of treatment.[6]

Nursing concerns

Caring for a child with a rare genetic disorder often has profound effects on health professionals and families.

i. Substantial knowledge basis for nursing practice.
ii. Critically analyzes the history and physical assessment findings.
iii. Collaboration.
iv. Coordination of Care.
v. Genetic counseling.
vi. Health Teaching and Health Promotion.
vii. Family support and parental involvement in care.
viii. Close monitoring and avoidance of complications.
ix. Facilitates referrals.
x. Support group.
xi. Research.

xii. Resource Utilization.

xiii. Leadership.

Conclusion

Recognition of hyperekplexia in the neonatal period is essential in avoiding misdiagnoses like epilepsy. Association with recurrent apnoeic spells, cardiac arrhythmia, SIDS, and severe feeding difficulties with apnoea indicates the need for providing home monitoring and appropriate parental counseling.[7,8]

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Conflict of interest

The author declares no conflict of interest.

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